20191003_data_release_notes

Overview of changes in the release of October 3, 2019

Please also see our RSS feed for information about accessing the data.

Overview of submissions: 2019

759562
778673
782638
787656
795045
811551
819827
825177
881419
888298

Content

Brief	Explanation
Laboratory for Molecular Medicine, Partners HealthCare Personalized Medicine	Laboratory for Molecular Medicine, Partners HealthCare Personalized Medicine submitted updates to 9,922 records.
Lineagen Inc.	Lineagen Inc. submitted 4,443 novel variant interpretations.
Wong Mito Lab, Molecular and Human Genetics, Baylor College of Medicine	Wong Mito Lab, Molecular and Human Genetics, Baylor College of Medicine submitted 550 novel interpretations for variants in mitochondrial tRNA genes.
Molecular Diagnostic Laboratory for Inherited Cardiovascular Disease, Montreal Heart Institute	Molecular Diagnostic Laboratory for Inherited Cardiovascular Disease, Montreal Heart Institute submitted 381 novel variant interpretations.
GenomeConnect, ClinGen	GenomeConnect, ClinGen submitted 260 novel variant interpretations and updates to 4 records.
Equipe Genetique des Anomalies du Developpement, Université de Bourgogne	Equipe Genetique des Anomalies du Developpement, Université de Bourgogne submitted 220 novel variant interpretations.
Yale Center for Mendelian Genomics, Yale University	Yale Center for Mendelian Genomics, Yale University submitted 137 novel variant interpretations.
Department of Pediatrics, Gifu University	Department of Pediatrics, Gifu University submitted 106 novel variant interpretations.
use of MANE transcripts for preferred names for variants in ClinVar	We calculate a preferred name for each variant in ClinVar, often using a RefSeq transcript as the reference sequence. We are in the process of preferentially using transcripts from the MANE project in the preferred name.
	For more information about MANE transcripts, read this NCBI blog post:
	https://ncbiinsights.ncbi.nlm.nih.gov/2019/03/12/mane-select-v0-5/
	and RefSeq's documentation:
	https://www.ncbi.nlm.nih.gov/refseq/MANE/
missing data for GRCh37 to be restored	The VCF files for GRCh37 posted at 10/2/19 ~10:15 am are missing some data for GRCh37 and for mitochondrial variants, as in last week's release. We are generating corrected files and will post them in a few hours.
	UPDATE: Updated VCF files were posted at 10/3/19, 5:55 pm. We have restored most of the missing data; more fixes will be in place for next week's release.

Overview of submissions: 2018

Jan 01, 2018	579543
Feb 01, 2018	582113
Mar 01, 2018	593651
Apr 01, 2018	610005
May 01, 2018	645149
Jun 01, 2018	676018
Jul 01, 2018	676575
Aug 01, 2018	685942
Sep 01, 2018	701880
Oct 01, 2018	708726
Nov 01, 2018	715516
Dec 01, 2018	749203

Overview of submissions: 2017

396005
405182
406220
446265
482941
486420
488658
492592
504299
512373
517157
519359

Overview of submissions: 2016

Jan 01, 2016	172867
Feb 01, 2016	176710
Mar 01, 2016	178032
Apr 01, 2016	180549
May 01, 2016	181155
Jun 01, 2016	192617
Jul 01, 2016	204415
Aug 01, 2016	209842

Sep 01, 2016	210200
Oct 01, 2016	213499
Nov 01, 2016	236420
Dec 01, 2016	240042

Overview of submissions: 2015

149013
156999
162455
171408
172044
173236
184506
154686
158580
160538
170931
172006

Overview of submissions: 2014

Jan 01, 2014	68204
Feb 01, 2014	73492
Mar 01, 2014	83343
Apr 01, 2014	111501
May 01, 2014	112349
Jun 01, 2014	117209
Jul 01, 2014	127132
Aug 01, 2014	127557
Sep 1, 2014	143114
Oct 1, 2014	143601
Nov 1, 2014	144117
Dec 1, 2014	148008

Overview of Submissions: 2013

Apr 05, 2013	30333
May 01, 2013	30386
Jun 01, 2013	39047
Jul 01, 2013	39170

Sep 01, 2013 50263
36p 01, 2013 30203
Oct 01, 2013 52047
Nov 01, 2013 64750
Dec 01, 2013 64881